



## DHODH gene

dihydroorotate dehydrogenase (quinone)

### Normal Function

The *DHODH* gene provides instructions for making an enzyme called dihydroorotate dehydrogenase. This enzyme is involved in producing pyrimidines, which are building blocks of DNA, its chemical cousin RNA, and molecules such as ATP and GTP that serve as energy sources in the cell. Dihydroorotate dehydrogenase functions within mitochondria, the energy-producing centers within cells. Specifically, this enzyme converts a molecule called dihydroorotate to a molecule called orotic acid. In subsequent steps, other enzymes modify orotic acid to produce pyrimidines.

### Health Conditions Related to Genetic Changes

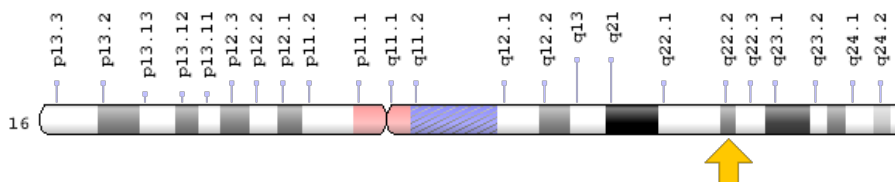
#### Miller syndrome

At least 11 mutations in the *DHODH* gene have been found to cause Miller syndrome. Most of these mutations change single protein building blocks (amino acids) in dihydroorotate dehydrogenase, which likely impairs the enzyme's ability to function normally. It is unclear exactly how *DHODH* gene mutations lead to the signs and symptoms of Miller syndrome.

### Chromosomal Location

Cytogenetic Location: 16q22.2, which is the long (q) arm of chromosome 16 at position 22.2

Molecular Location: base pairs 72,008,744 to 72,027,659 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- DHODEHase
- dihydroorotate dehydrogenase
- dihydroorotate dehydrogenase, mitochondrial
- dihydroorotate dehydrogenase, mitochondrial precursor
- dihydroorotate oxidase
- human complement of yeast URA1
- POADS
- PYRD\_HUMAN
- URA1

## Additional Information & Resources

### Educational Resources

- Biochemistry (fifth edition, 2002): Orotate Acquires a Ribose Ring from PRPP to Form a Pyrimidine Nucleotide and Is Converted into Uridylate  
<https://www.ncbi.nlm.nih.gov/books/NBK22447/#A3485>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DHODH%5BTIAB%5D%29+OR+%28dihydroorotate+dehydrogenase%5BTIAB%5D%29%29+OR+%28%28POADS%5BTIAB%5D%29+OR+%28DHODEhase%5BTIAB%5D%29+OR+%28dihydroorotate+oxidase%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- DIHYDROOROTATE DEHYDROGENASE  
<http://omim.org/entry/126064>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_DHODH.html](http://atlasgeneticsoncology.org/Genes/GC_DHODH.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=DHODH%5Bgene%5D>

- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=2867](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2867)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/1723>
- UniProt  
<http://www.uniprot.org/uniprot/Q02127>

## Sources for This Summary

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